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Abstract Title:- Optical Genome Mapping in Structural Variant Identification

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Aims:-The chromosomal translocations, deletions, duplications, inversions, and repeat expansions are called Structural Variants (SVs).Usually detecting SVs requires multiple techniques. For example, chromosomal microarrays (CMA) can detect large deletions and duplications but cannot identify balanced reciprocal translocations, repeat expansion, SVs smaller than 40 kb, genomic location, and orientation of copy number gains. The breakthrough in sequencing technologies identifies the disease-causing variants in single gene disorders by short-read sequencing. However, the detection of SVs still remains challenging due to the short read length and repetitive nature. Although long-read sequencing is a better choice, the error rate is high and the pipelines are difficult to establish. Apparently, recent advances in next-generation cytogenomic tools like optical genome mapping (OGM) have found a way to detect all types of SVs. We present five cases analyzed using OGM technique

Methods:- Optical Genome Mapping, OGM is a high-resolution technology that uses ultra-long DNA that is fluorescently labelled at hexamer motifs found throughout the genome which creates a barcode pattern similar to G-bands making way to detect all SVs at high resolution by comparing to a reference genome. We have used this technology for five cases and we present our results.

Results:- Five cases of SVs were analyzed by de novo assembly pipeline and Bionano Access v1.7X software. All the variants are represented in a Circos plot which can be easily analyzed. Further, long-range PCR followed by amplicon sequencing was performed to delineate the breakpoints till the base pair level. In our study, we showed that OGM is capable of detecting all types of SVs.

Conclusions:- To conclude optical genome mapping can be used as an alternative approach for Karyotyping, FISH, and Microarrays due to its ease of data analysis which will be discussed.

Keywords:- Optical Genome Mapping, Structural Variants, Cytogenomics, OGM, Bionano